

## GEPHE SUMMARY

<p>Kit (type III receptor protein-tyrosine kinase) (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>GP00000518</p> <p>Entry Status</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a>)</p> <p>Coloration (coat) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title</a>)</p> <p>Felis catus</p> <p>Felis catus - Birman (white gloving)</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Felis catus</p> <p>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Felis+catus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Felis+catus^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>domestic cat</p> <p>Synonyms</p> <p>Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Felinae; Felis</p> <p>Parent</p> <p>Felis () - (Rank: genus)</p> <p>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682</a>)</p> <p>NCBI Taxonomy ID</p> <p>9685</p> <p>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Felis catus</p> <p>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Felis+catus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Felis+catus^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>domestic cat</p> <p>Synonyms</p> <p>Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Felinae; Felis</p> <p>Parent</p> <p>Felis () - (Rank: genus)</p> <p>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682</a>)</p> <p>NCBI Taxonomy ID</p> <p>9685</p> <p>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Taxon B Description</p> <p>Felis catus - Birman (white gloving)</p>
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## GENOTYPIC CHANGE

<p>Kit</p> <p>W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfscow3; Sl</p> <p>10090.ENSMUSP00000005815</p> <p>(<a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815</a>)</p> <p>Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.</p> <p>GO:0004888 : transmembrane signaling receptor activity</p> <p>(<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004888">https://www.ebi.ac.uk/QuickGO/term/GO:0004888</a>)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P05532 (<a href="http://www.uniprot.org/uniprot/P05532">http://www.uniprot.org/uniprot/P05532</a>)</p> <p>()</p> <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
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GO:0005524 : ATP binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005524>)  
GO:0042803 : protein homodimerization activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)  
GO:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)  
GO:0002020 : protease binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0002020>)  
GO:0004714 : transmembrane receptor protein tyrosine kinase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004714>)  
GO:0004713 : protein tyrosine kinase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004713>)  
GO:0019955 : cytokine binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019955>)  
GO:0005020 : stem cell factor receptor activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005020>)

#### GO - Biological Process

GO:0043066 : negative regulation of apoptotic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)  
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)  
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)  
GO:0070374 : positive regulation of ERK1 and ERK2 cascade  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)  
GO:0035234 : ectopic germ cell programmed cell death  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035234>)  
GO:0035162 : embryonic hemopoiesis  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035162>)  
GO:0008584 : male gonad development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008584>)  
GO:0001541 : ovarian follicle development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001541>)  
GO:0008284 : positive regulation of cell proliferation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)  
GO:0043406 : positive regulation of MAP kinase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043406>)  
GO:0010628 : positive regulation of gene expression  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)  
GO:0043410 : positive regulation of MAPK cascade  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043410>)  
GO:0007283 : spermatogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0007283>)  
GO:0008360 : regulation of cell shape  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008360>)  
GO:0048070 : regulation of developmental pigmentation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048070>)  
GO:0006468 : protein phosphorylation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006468>)  
GO:0060326 : cell chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060326>)  
GO:0006935 : chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0006935>)  
GO:0048565 : digestive tract development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048565>)  
GO:0006954 : inflammatory response  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006954>)  
GO:0019221 : cytokine-mediated signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019221>)  
GO:0048863 : stem cell differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048863>)  
GO:0048066 : developmental pigmentation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048066>)  
GO:0030318 : melanocyte differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)  
GO:0009968 : negative regulation of signal transduction  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009968>)  
GO:0046777 : protein autophosphorylation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046777>)  
GO:0030218 : erythrocyte differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030218>)  
GO:0018108 : peptidyl-tyrosine phosphorylation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018108>)  
GO:0097067 : cellular response to thyroid hormone stimulus  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097067>)  
GO:1904349 : positive regulation of small intestine smooth muscle contraction  
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904349>)  
GO:0000187 : activation of MAPK activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000187>)  
GO:0046427 : positive regulation of JAK-STAT cascade  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046427>)  
GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042531>)  
GO:0030335 : positive regulation of cell migration  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030335>)  
GO:0046686 : response to cadmium ion  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046686>)

GO:0035556 : intracellular signal transduction  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)  
GO:0031532 : actin cytoskeleton reorganization  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031532>)  
GO:0002371 : dendritic cell cytokine production  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002371>)  
GO:0050910 : detection of mechanical stimulus involved in sensory perception of sound  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050910>)  
GO:0050673 : epithelial cell proliferation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050673>)  
GO:0038162 : erythropoietin-mediated signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038162>)  
GO:0038093 : Fc receptor signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038093>)  
GO:0007281 : germ cell development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007281>)  
GO:0008354 : germ cell migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0008354>)  
GO:0006687 : glycosphingolipid metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006687>)  
GO:0035701 : hematopoietic stem cell migration  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035701>)  
GO:0030097 : hemopoiesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0030097>)  
GO:0002327 : immature B cell differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002327>)  
GO:0038109 : Kit signaling pathway (<https://www.ebi.ac.uk/QuickGO/term/GO:0038109>)  
GO:0030032 : lamellipodium assembly  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030032>)  
GO:0002320 : lymphoid progenitor cell differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002320>)  
GO:0002551 : mast cell chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0002551>)  
GO:0032762 : mast cell cytokine production  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032762>)  
GO:0043303 : mast cell degranulation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043303>)  
GO:0060374 : mast cell differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060374>)  
GO:0035855 : megakaryocyte development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035855>)  
GO:0097326 : melanocyte adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0097326>)  
GO:0097324 : melanocyte migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0097324>)  
GO:0002573 : myeloid leukocyte differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002573>)  
GO:0002318 : myeloid progenitor cell differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002318>)  
GO:0043069 : negative regulation of programmed cell death  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043069>)  
GO:1904343 : positive regulation of colon smooth muscle contraction  
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904343>)  
GO:0051091 : positive regulation of DNA-binding transcription factor activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051091>)  
GO:0048170 : positive regulation of long-term neuronal synaptic plasticity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048170>)  
GO:0045747 : positive regulation of Notch signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045747>)  
GO:0031274 : positive regulation of pseudopodium assembly  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031274>)  
GO:0120072 : positive regulation of pyloric antrum smooth muscle contraction  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0120072>)  
GO:1905065 : positive regulation of vascular smooth muscle cell differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:1905065>)  
GO:1904251 : regulation of bile acid metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904251>)  
GO:0009314 : response to radiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0009314>)  
GO:0048103 : somatic stem cell division  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048103>)  
GO:0035019 : somatic stem cell population maintenance  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035019>)  
GO:0007286 : spermatid development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007286>)  
GO:0030217 : T cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030217>)  
GO:0043586 : tongue development (<https://www.ebi.ac.uk/QuickGO/term/GO:0043586>)  
GO:0008542 : visual learning (<https://www.ebi.ac.uk/QuickGO/term/GO:0008542>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)  
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)  
GO:0005887 : integral component of plasma membrane  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)  
GO:0043235 : receptor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0043235>)

GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)  
GO:0009986 : cell surface (<https://www.ebi.ac.uk/QuickGO/term/GO:0009986>)  
GO:0009898 : cytoplasmic side of plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0009898>)  
GO:0009897 : external side of plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0009897>)  
GO:0001669 : acrosomal vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0001669>)  
GO:0005911 : cell-cell junction (<https://www.ebi.ac.uk/QuickGO/term/GO:0005911>)  
GO:0042629 : mast cell granule (<https://www.ebi.ac.uk/QuickGO/term/GO:0042629>)

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No^"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding^"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=))

Aberration Type

Indel ([https://www.gephebase.org/search-criteria?/and+Aberration Type="+Indel^"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=))

Indel Size

1-9 bp

Molecular Details of the Mutation

Two nucleotide changes affecting site 3 of codon 345 and site 1 of codon 346 - resulting in Glu345Asp + His346Asn  
c. 1035\_1036delinsCA GAGCAY > GACAAY

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Association Mapping^"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))

Main Reference

Comparative analysis of the domestic cat genome reveals genetic signatures underlying feline biology and domestication. (2014) (<https://pubmed.ncbi.nlm.nih.gov/25385592>)

Authors

Montague MJ; Li G; Gandolfi B; Khan R; Aken BL; Searle SM; Minx P; Hillier LW; Koboldt DC; Davis BW; Driscoll CA; Barr CS; Blackstone K; Quilez J; Lorente-Galdos B; Marques-Bonet T; Alkan C; Thomas GW; Hahn MW; Menotti-Raymond M; O'Brien SJ; Wilson RK; Lyons LA; Murphy WJ; Warren WC

Abstract

Little is known about the genetic changes that distinguish domestic cat populations from their wild progenitors. Here we describe a high-quality domestic cat reference genome assembly and comparative inferences made with other cat breeds, wildcats, and other mammals. Based upon these comparisons, we identified positively selected genes enriched for genes involved in lipid metabolism that underpin adaptations to a hypercarnivorous diet. We also found positive selection signals within genes underlying sensory processes, especially those affecting vision and hearing in the carnivore lineage. We observed an evolutionary tradeoff between functional olfactory and vomeronasal receptor gene repertoires in the cat and dog genomes, with an expansion of the feline chemosensory system for detecting pheromones at the expense of odorant detection. Genomic regions harboring signatures of natural selection that distinguish domestic cats from their wild congeners are enriched in neural crest-related genes associated with behavior and reward in mouse models, as predicted by the domestication syndrome hypothesis. Our description of a previously unidentified allele for the gloving pigmentation pattern found in the Birman breed supports the hypothesis that cat breeds experienced strong selection on specific mutations drawn from random bred populations. Collectively, these findings provide insight into how the process of domestication altered the ancestral wildcat genome and build a resource for future disease mapping and phylogenomic studies across all members of the Felidae.

Additional References

## RELATED GEPHE

Related Genes

6 (Agouti, MC1R, Melanophilin (MLPH), Taqpep, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID="+9685^"/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=))

Related Haplotypes

2 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase="+Kit \(type III receptor protein-tyrosine kinase\)^"/and+Taxon ID="+9685^"/or+Gene Gephebase="+Kit \(type III receptor protein-tyrosine kinase\)^"/and+Taxon ID="+9685^"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=))

## EXTERNAL LINKS

## COMMENTS

cis-regulatory contribution is not excluded - The two coding mutations are adjacent. It looks like they both occurred all at once because no individual has been found with only one of the two mutations. @SeveralMutationsWithEffect @TwoAdjacentNucleotideChanges <https://omia.org/OMIA001580/9685/>